Ashford and St. Peter's Hospitals
NHS Foundation Trust

Downs Syndrome Guideline for Neonatal Unit

Author: Martins Aso

Supervisor: Samantha Edwards

Target Audience: Neonatal Doctors and Nurses, Midwives

Equality impact assessment: Neonatal Governance Group

Guideline History		
Date	**Comments**	**Approved By**
July 2021	Revised Guideline	NGG

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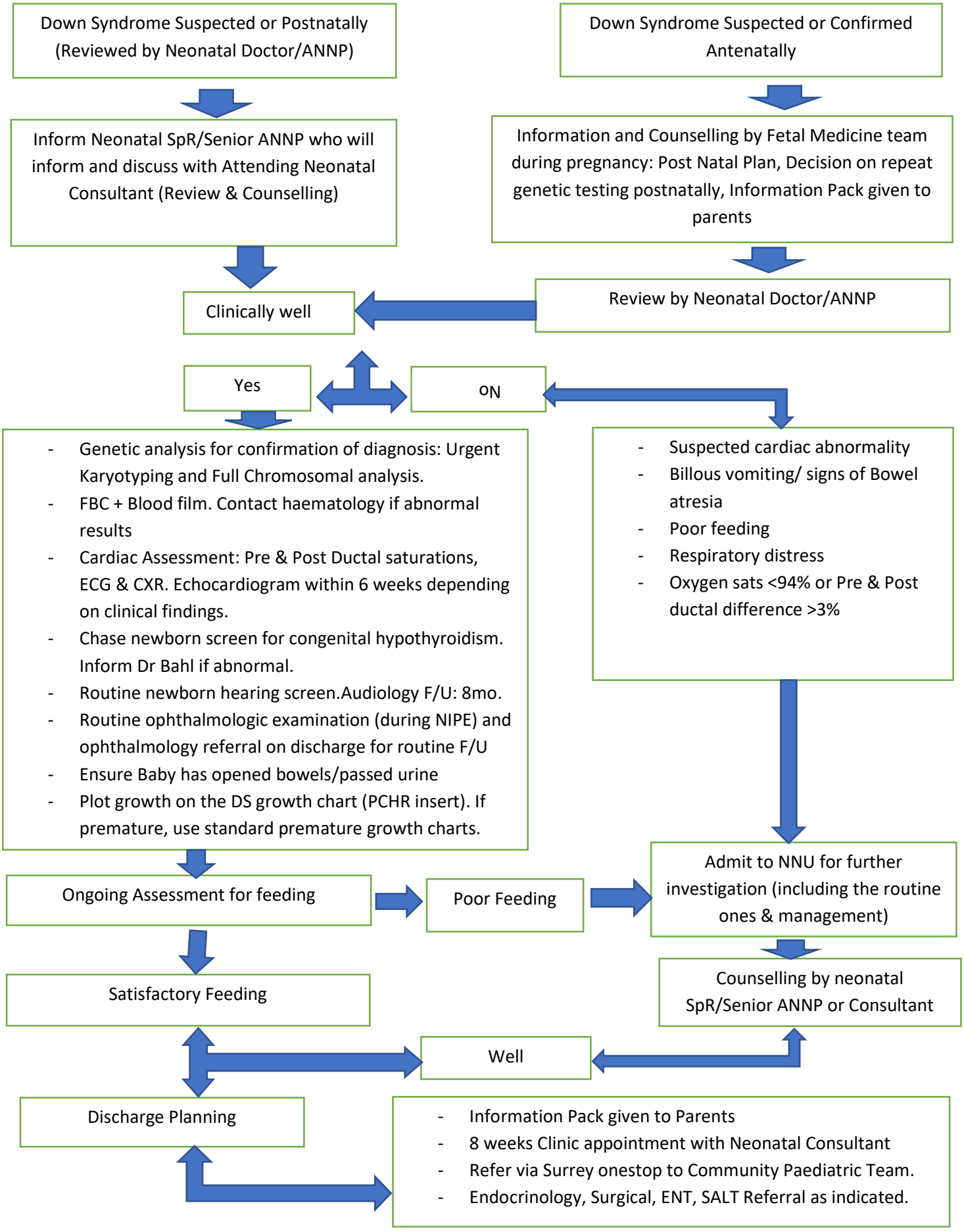
Purpose:

This guideline proposes the appropriate management pathway at each stage, from prenatal diagnosis and neonatal care.

This guideline is largely based on work done by the Down Syndrome Medical Interest Group (DSMIG, UK and Ireland) who have produced guidelines for basic medical surveillance in children with Down Syndrome. This guideline and a wide range of other health information, can be found at www.dsmig.org.uk.

NICU

Post Natal Management of Down Syndrome



Introduction

Down syndrome is a chromosomal abnormality that results from an extra copy of chromosome 21 leading to characteristic clinical features in the affected child.

Down syndrome accounts for 1:1000 live births in the UK.

Some of the features include: flattened face, microcephaly, short neck, protruding tongue, upward slanting palpebral fissures, unusually shaped or small ears, poor muscle tone, broad short hands with single palmar crease, relatively short fingers and small hands and feet, Brushfield spots (tiny white spots on the iris).

Neonates with Down syndrome are at an increased chance of a wide range of medical problems in the neonatal period. Based on current evidence the following good practice recommendations have been made to help guide clinical practice.

Antenatal Diagnosis
Some babies with Down syndrome are diagnosed antenatally and the responsibility for counselling lies with the local antenatal team.
Screening for Down Syndrome in the baby is done in the first trimester. The safe test is a non-invasive prenatal testing which screens for Down's, Edward's and Patau syndrome. Mother's blood sample is sent to the NHS laboratory at St Georges hospital for assessment. Results are available within 7 days of sample receipt. In order to confirm a diagnosis, genetic testing through chorionic villus sampling or early amniocentesis (18-22weeks) will be required. A repeat genetic testing will be required when the baby is born. NB: If the confirmatory test is done by a late Amniocentesis (third trimester), then a repeat genetic testing will not be required.

Postnatal Diagnosis
In cases where diagnosis has not been made antenatally and there is clinical suspicion of Down syndrome, please discuss with NICU attending consultant, so that a health professional with sufficient knowledge of Down syndrome can review the baby to confirm clinical diagnosis and meet with the parents to discuss the plan.
Blood should be sent for genetic analysis to confirm the diagnosis. (Samples to be taken in EDTA Bottles and Lithium Heparin Bottles. It is advisable to ensure the bottle is full to the mark and sent to the Genetics Lab at Guys and St Thomas Hospital through the Lab at St Peters Hospital.) Results are usually available after 72hrs of sample receipt.
Blood should also be taken at the same time for FBC and blood film.

Action following Confirmed Diagnosis
Where a Confirmatory result is positive, parents should be told of the diagnosis as soon as possible, preferably by an appropriate experienced member of the neonatal team.
An information pack should be given to the parents. This may include:

- -The Down syndrome PCHR insert
- - Information on the relevant Down's Syndrome Association and their New Parent Guide

- Information about local support groups
- Down's Syndrome Heart Group

Parents should be sign posted to the NICU Website: www.ashfordstpeters.nhs.uk/leaflets-nicu for the necessary information. Get the Down syndrome PCHR insert from the neonatal community office.

The obstetrician, midwife, GP and HV should also be informed as soon as possible.

Post Natal Management of the Down Syndrome Patient

Routine neonatal examination with particular attention to common complications of Down syndrome such as bowel atresias, Hirschsprung's, cardiac defects and cataracts.

Cardiology

Between 40 and 60% of babies with Down syndrome have congenital heart defects. Of these 30 - 40% are complete atrioventricular septal defects (AVSD). Most AVSD can be successfully treated if the diagnosis is made early and the baby referred for full corrective surgery before irreversible pulmonary vascular disease (PVD) is established.

The attending Clinician should review the Fetal ECHO as well as the post natal plan if any and the report should be incorporated in the baby's record.
All babies should have Pre and Post Ductal saturations, ECG and CXR, then Echocardiogram within 6weeks or as inpatient depending on ECG/CXR findings. Discuss with the attending consultant regarding appropriate timing of scan.

If there is a heart defect, offer to refer the family to the Down's Heart Group which provides parent support for any child with a heart problem.

Haematology

If a diagnosis of Down syndrome is made antenatally, delayed clamping of the cord should still be performed, but limited to 1 minute only.

All babies with suspected or confirmed Down syndrome should have a FBC and a Blood film taken in the first 2-3 days of life, ideally at the same time as their genetic tests. Particular attention should be given to the peripheral blood blast percentage assessed by a haematologist with experience at reviewing neonatal blood films.

Transient Abnormal Myelopoiesis (TAM) is a congenital leukaemia unique to neonates with Down syndrome or mosaic trisomy 21. Previous names include Transient Myeloproliferative Disorder (TMD) and Transient Leukaemia of Down syndrome (TL-DS).

Any neonate with a blast percentage of >10% and/or clinical features suggestive of TAM should be discussed urgently with the regional paediatric haematology centre (GOSH) and a peripheral blood sample should be sent for *GATA1* mutation analysis in an accredited laboratory.
Any child who did not have a peripheral blood blast cell percentage performed in the first 3 days of life or in whom there was significant intra-uterine growth retardation (when blast

counts may be supressed) should be considered to be still at risk of clinical problems of TAM in the first 4-8 weeks of life and should be monitored accordingly. GATA1 mutation analysis should be considered.

If TAM is associated with clinical symptoms such as bruises, bleeding easily, liver & Renal impairment, infection; the neonate should be monitored closely until there is spontaneous resolution of symptoms and thereafter with a FBC and blood film 3 monthly until the age of 2 years and then 6 monthly until the age of 4 years. Abnormal blood counts or blood film appearance should prompt early investigation.

If TAM is diagnosed and the neonate is asymptomatic then a FBC and blood film should be monitored 3 monthly until the age of 2 years and if they are normal then 6 monthly until 4 years of age.

Thyroid Function
The incidence of congenital hypothyroidism in Down syndrome is more common and occurs in 1-3.6% of children.
The newborn screening programme for congenital hypothyroidism based on TSH analysis is satisfactory. Please chase the result and if abnormal, discuss with Dr Bahl and follow the hypothyroidism guideline. Contact St Helier Hospital laboratory on 02082962991 or email est-tr.swtnewbornscreening@nhs.net for results.

Hearing
Routine newborn hearing screen should be done for all newborn babies with Down syndrome.
Otitis media with effusion (glue ear) affects up to 35% of children with Down syndrome at birth. There is a higher incidence of ossicular abnormalities in Down syndrome which may present with a conductive hearing loss.
Audiology follow-up should be arranged for 8 months, even if screening is normal.

Vision
All babies with Down syndrome should have routine ophthalmologic examination by an experienced member of the team during NIPE and this should be documented. Refer any possible abnormalities immediately to an ophthalmologist but if normal examination, Baby should have routine ophthalmologic follow up booked at discharge.
Babies with Down syndrome have a tenfold increase in congenital cataract and infantile glaucoma.

Digestive system
Approximately 7% of children with Down syndrome have congenital malformations of the GIT. Defects include duodenal stenosis/atresia (3.9%), hirschprung's disease (2.6%), anal stenosis/atresia (1.0%), oesophageal atresia/tracheoesophageal fistula (0.4%) and pyloric stenosis (0.3%). Its important to ensure bowels are open within 24hours.
Some babies with Down syndrome may have difficulties establishing feeds. Thus, help and support should be given to mothers who wish to breastfeed as well as supporting those who

want to bottle feed. A nasogastric tube may be needed to supplement feeds, whilst breastfeeding is being established.
Consider an early referral to the infant feeding team, SALT and dietician; especially if the baby is admitted to the neonatal unit.

Renals
Several congenital abnormalities of the renal and urinary tract have been reported in children with Down syndrome. These include renal hypoplasia, obstructive uropathy including posterior urethral valves, glomerular microcysts, hypospadias and undescended testicles. Some of these abnormalities may be picked up on antenatal scans and should be dealt with as appropriate. Check that the antenatal scans of the renal tract were reported as normal and that the baby passes urine normally. If in any doubt request a renal tract scan.

Discharge Planning

- Outpatient appointment 8weeks with Neonatal Consultant.
- Plot growth on the Down Syndrome growth chart (PCHR insert) (if born prematurely, use standard premature growth charts) and add to parent held red book.
- Refer via Surrey onestop to Community Paediatric Team. See Link below: https://childrenshealthsurrey.nhs.uk/services/one-stop
- Offer further information to Parents (Down Syndrome Association https://www.downs-syndrome.org.uk; https://www.nhs.uk/conditions/downs-syndrome/)
- Specialist referral if needed: Cardiology, Haematology, Surgical, ENT (if concerns about upper airway), SALT (If feeding difficulties).

Referral to Community Paediatric Team

- This referral is done through Surrey OneStop and should include a request for the baby to be seen by community physiotherapist, SALT, and community paediatrician for a multi-disciplinary review
- Need to have consent from parents (verbal), and their email address/phone number for the online referral
- Please copy the reference number in baby's medical records (and BADGER).
- Please enter the details of the named neonatal consultant for the baby on the one-stop referral form, to enable smooth continuation of care after discharge.

References

1. www.dsmig.org.uk. Down Syndrome Medical Interest Group. DSMIG Best Practical Guidance. September 2018.
2. Nottingham Down Syndrome guidelines 2017.

4. Guideline Governance

a. Scope

This guideline in relevant to all staff caring for babies across neonatal intensive care, transitional care and maternity.

b. Purpose

i. This guidelines aims to facilitate a common approach to the management of babies admitted under neonatal care. At times deviation from the guideline may be necessary, this should be documented and is the responsibility of the attending consultant.

ii. This guideline is subject to regular review to ensure ongoing evidence based practice.

c. Duties and Responsibilities

All health care professionals responsible for the care of newborn babies should be familiar with the guidance in this document, and ensure ongoing training.

d. Approval and Ratification

This guideline will be approved and ratified by the Neonatal Guidelines Group.

e. Dissemination and Implementation

i. This guideline will be uploaded to the trust intranet 'Neonatal Guidelines' page and thus available for common use.
ii. This guideline will be shared as part of ongoing education within the Neonatal Unit for both medical and nursing staff.
iii. All members of staff are invited to attend and give comments on the guideline as part of the ratification process.

f. Review and Revision Arrangements

a. This policy will be reviewed on a 5 yearly basis.
b. If new information comes to light prior to the review date, an earlier review will be prompted.
c. Amendments to the document shall be clearly marked on the document control sheet and the updated version uploaded to the intranet. Minor amendments will be ratified through the Neonatal Guidelines Group. A minor amendment would consist of no major change in process, and includes but is not limited to, amendments to documents within the appendices.

g. Equality Impact Assessment

Background • Who was involved in the Equality Impact Assessment
Guidelines chair
Methodology • A brief account of how the likely effects of the policy was assessed (to include race and ethnic origin, disability, gender, culture, religion or belief, sexual orientation, age) • The data sources and any other information used • The consultation that was carried out (who, why and how?)
All groups of staff and patients considered
Key Findings • Describe the results of the assessment • Identify if there is adverse or a potentially adverse impacts for any equalities groups
No evidence of discrimination
Conclusion • Provide a summary of the overall conclusions
No evidence of discrimination
Recommendations • State recommended changes to the proposed policy as a result of the impact assessment • Where it has not been possible to amend the policy, provide the detail of any actions that have been identified • Describe the plans for reviewing the assessment
Appropriate for general use

h. Document Checklist

To be completed (electronically) and attached to any document which guides practice when submitted to the appropriate committee for approval or ratification.

Title of the document:

Policy (document) Author:

Executive Director:

		Yes/No/ Unsure/NA	Comments
1.	**Title**		
	Is the title clear and unambiguous?	**Y**	
	Is it clear whether the document is a guideline, policy, protocol or standard?	**Y**	
2.	**Scope/Purpose**		
	Is the target population clear and unambiguous?	**Y**	
	Is the purpose of the document clear?	**Y**	
	Are the intended outcomes described?	**Y**	
	Are the statements clear and unambiguous?	**Y**	
3.	**Development Process**		
	Is there evidence of engagement with stakeholders and users?	**Y**	
	Who was engaged in a review of the document (list committees/ individuals)?	**Y**	
	Has the policy template been followed (i.e. is the format correct)?	**Y**	
4.	**Evidence Base**		
	Is the type of evidence to support the document identified explicitly?	**Y**	
	Are local/organisational supporting documents referenced?	**Y**	
5.	**Approval**		
	Does the document identify which committee/group will approve/ratify it?	**Y**	
	If appropriate, have the joint human resources/staff side committee (or equivalent) approved the document?	**Y**	
6.	**Dissemination and Implementation**		
	Is there an outline/plan to identify how this will be done?	**Y**	
	Does the plan include the necessary training/support to ensure compliance?	**Y**	
7.	**Process for Monitoring Compliance**		

		Yes/No/ Unsure/NA	Comments
	Are there measurable standards or KPIs to support monitoring compliance of the document?		
8.	**Review Date**		
	Is the review date identified and is this acceptable?	**Y**	
9.	**Overall Responsibility for the Document**		
	Is it clear who will be responsible for coordinating the dissemination, implementation and review of the documentation?	**Y**	
10.	**Equality Impact Assessment (EIA)**		
	Has a suitable EIA been completed?	Y	

Committee Approval (Neonatal Guidelines Committee)			
If the committee is happy to approve this document, please complete the section below, date it and return it to the Policy (document) Owner			
Name of Chair	**S. Edwards**	**Date**	**21 August 2021**
Ratification by Management Executive (if appropriate)			
If the Management Executive is happy to ratify this document, please complete the date of ratification below and advise the Policy (document) Owner			
Date: n/a			